1: Atherosclerosis 1989 Sep;79(1):85-91

DNA polymorphisms at the lipoprotein lipase gene: associations in normal and hypertriglyceridaemic subjects.

Chamberlain JC, Thorn JA, Oka K, Galton DJ, Stocks J

Diabetes and Lipid Laboratory, St. Bartholomew's Hospital, London, U.K.

Lipoprotein lipase is a rate determining enzyme for the removal of triglyceride-rich lipoproteins from the blood stream. We examined whether genetic variation at the lipoprotein lipase gene locus was related to the fasting plasma level of triglycerides in both a normal and hypertriglyceridaemic population. Two restriction fragment length polymorphisms revealed by the enzymes PvuII and HindIII generated alleles designated H1, 17.5 kb;H2, 8.7 kb;P1, 7.0 kb;P2, 4.4 and 2.5 kb, respectively. These were studied in 46 Caucasian hypertriglyceridaemic subjects in comparison with 86 normolipidaemic controls. The respective allelic frequencies were H1 0.211, H2 0.789 and H1 0.414, H2 0.586 (p less than 0.01). Similar differences in allelic frequencies were found in a smaller group of Japanese hypertriglyceridaemic subjects (n = 29) compared to Japanese controls (n = 41, p less than 0.01). Ninety-three healthy Caucasians were genotyped for both polymorphic sites to relate to levels of plasma triglyceride. We found that individuals with genotype P1P1 had fasting triglyceride levels of 0.96 +/- 0.31 mmol/l (n = 20) compared to genotype P2P2 with levels of 1.31 +/- 0.66 mmol/l (n = 30, p less than 0.02); heterozygous subjects (P1P2) had intermediate levels of plasma triglyceride (1.15 +/- 0.46 mmol/l, n = 43). The HindIII alleles were not significantly associated with variation in levels of plasma triglyceride, cholesterol, or HDL-cholesterol. We conclude that DNA variations at, or around, the lipoprotein lipase gene may constitute genetic determinants for both the population variation in plasma triglyceride levels as well as for the common metabolic disorder of primary hypertriglyceridaemia.

PMID: 2803349, UI: 90026498